PABPN1 gene

poly(A) binding protein nuclear 1

Normal Function

The *PABPN1* gene provides instructions for making a protein that is active (expressed) throughout the body. In cells, the PABPN1 protein plays an important role in processing molecules called messenger RNAs (mRNAs), which serve as genetic blueprints for making proteins. The PABPN1 protein attaches (binds) to the end of an mRNA molecule at a region called the polyadenine tail or poly(A) tail. Poly(A) tails consist of many adenine molecules, one of the building blocks of RNA and its chemical cousin, DNA. Poly(A) tails are needed to protect the mRNA from being broken down and allow the mRNA to move within cells. The PABPN1 protein helps add adenines to the poly(A) tail through a process called polyadenylation. The PABPN1 protein may also be involved in regulating mRNA production.

The PABPN1 protein contains an area where the protein building block (amino acid) alanine is repeated 10 times. This stretch of alanines is known as a polyalanine tract. The role of the polyalanine tract in normal PABPN1 protein function is unknown.

Health Conditions Related to Genetic Changes

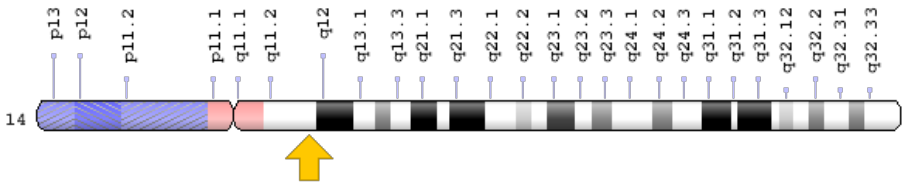
oculopharyngeal muscular dystrophy

At least 10 different mutations in the *PABPN1* gene have been found to cause oculopharyngeal muscular dystrophy. All of these mutations result in a PABPN1 protein with an expanded polyalanine tract that is 11 to 17 alanines long. The extra alanines cause the PABPN1 protein to form clumps within muscle cells that are thought to impair the normal functioning of muscle cells and eventually cause cell death. The progressive loss of muscle cells most likely causes the muscle weakness seen in people with oculopharyngeal muscular dystrophy.

Chromosomal Location

Cytogenetic Location: 14q11.2, which is the long (q) arm of chromosome 14 at position 11.2

Molecular Location: base pairs 23,320,188 to 23,326,185 on chromosome 14 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- OPMD
- PAB2
- PABP2
- PABP2_HUMAN
- poly(A) binding protein 2
- poly(A) binding protein II
- poly(A) binding protein, nuclear 1

Additional Information & Resources

Educational Resources

- The Cell: A Molecular Approach (second edition, 2002): Processing of mRNA in Eukaryotes
  https://www.ncbi.nlm.nih.gov/books/NBK9864/#A1031

GeneReviews

- Oculopharyngeal Muscular Dystrophy
  https://www.ncbi.nlm.nih.gov/books/NBK1126

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28PABPN1%5BTIAB%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D

OMIM

- POLYADENYLATE-BINDING PROTEIN, NUCLEAR, 1
  http://omim.org/entry/602279

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_PABPN1.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=PABPN1%5Bgene%5D
- HGNC Gene Family: RNA binding motif containing
  http://www.genenames.org/cgi-bin/genefamilies/set/725
- HGNC Gene Symbol Report
  http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=8565
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/8106
- UniProt
  http://www.uniprot.org/uniprot/Q86U42

Sources for This Summary

- Kühn U, Wahle E. Structure and function of poly(A) binding proteins. Biochim Biophys Acta. 2004 May 25;1678(2-3):67-84. Review.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/15157733
- Marie-Josée Sasseville A, Caron AW, Bourget L, Klein AF, Dicaire MJ, Rouleau GA, Massie B, Langelier Y, Brais B. The dynamism of PABPN1 nuclear inclusions during the cell cycle. Neurobiol Dis. 2006 Sep;23(3):621-9. Epub 2006 Jul 24.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/16860991
- Müller T, Deschauer M, Kolbe-Fehr F, Zierz S. Genetic heterogeneity in 30 German patients with oculopharyngeal muscular dystrophy. J Neurol. 2006 Jul;253(7):892-5. Epub 2006 Apr 20.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/16619122
- OMIM: POLYADENYLATE-BINDING PROTEIN, NUCLEAR, 1
  http://omim.org/entry/602279

- Robinson DO, Hammans SR, Read SP, Sillibourne J. Oculopharyngeal muscular dystrophy (OPMD): analysis of the PABPN1 gene expansion sequence in 86 patients reveals 13 different expansion types and further evidence for unequal recombination as the mutational mechanism. Hum Genet. 2005 Mar;116(4):267-71. Epub 2005 Jan 12.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/15645184
- Robinson DO, Wills AJ, Hammans SR, Read SP, Sillibourne J. Oculopharyngeal muscular dystrophy: a point mutation which mimics the effect of the PABPN1 gene triplet repeat expansion mutation. J Med Genet. 2006 May;43(5):e23.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/16648376
  Free article on PubMed Central: https://www.ncbi.nlm.nih.gov/pmc/articles/PMC2564528/

Reprinted from Genetics Home Reference:
https://ghr.nlm.nih.gov/gene/PABPN1

Reviewed: December 2008
Published: March 21, 2017

Lister Hill National Center for Biomedical Communications
U.S. National Library of Medicine
National Institutes of Health
Department of Health & Human Services